

(19) World Intellectual Property
Organization
International Bureau



(43) International Publication Date
13 January 2005 (13.01.2005)

PCT

(10) International Publication Number
WO 2005/003390 A2

- (51) International Patent Classification⁷: **C12Q 1/68**
- (21) International Application Number:
PCT/IB2004/002435
- (22) International Filing Date: 2 July 2004 (02.07.2004)
- (25) Filing Language: English
- (26) Publication Language: English
- (30) Priority Data:
03291624.9 2 July 2003 (02.07.2003) EP
- (71) Applicant (for all designated States except US): **CON-SORTIUM NATIONAL DE RECHERCHE EN GENOMIQUE (CNRG)** [FR/FR]; 2, rue Gaston Crémieux, F-91000 Evry (FR).
- (72) Inventors; and
- (75) Inventors/Applicants (for US only): **GUT, Ivo, Glynne** [GB/FR]; 18, rue du Moulin Vert, F-75014 Paris (FR). **MAUGER, Florence** [FR/FR]; 21, Place Gauvain, F-91000 Evry (FR).
- (74) Agents: **MARTIN, Jean-Jacques** et al.; Cabinet Regimbeau, 20, rue de Chazelles, F-75847 Paris Cedex 17 (FR).
- (81) Designated States (unless otherwise indicated, for every kind of national protection available): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NA, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.
- (84) Designated States (unless otherwise indicated, for every kind of regional protection available): ARIPO (BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PL, PT, RO, SE, SI, SK, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).
- Declaration under Rule 4.17:**
— of inventorship (Rule 4.17(iv)) for US only
- Published:**
— without international search report and to be republished upon receipt of that report
- For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.*

(54) Title: METHOD FOR DETECTION OF MUTATIONS IN DNA

(57) Abstract: A method for the detection in a given DNA sequence of DNA mutations, single nucleotide polymorphisms, and insertions and deletions, said method comprising the step of a) producing replicate(s) with an engineered polymerase of said given DNA sequence with part or all of at least one of the four natural DNA bases exchanged against a not natural base; b) using said not natural base to cleave the replicate(s) obtained in step a) and to produce a DNA product presenting sequence-specific fragments; c) analyzing said sequence-specific fragments obtained in step b) by mass spectrometry to get sequence-specific fragment patterns; and d) using the sequence-specific fragment patterns obtained in step c) to identify sequence changes relative to a reference to said given DNA sequence. A kit for the detection of DNA mutations, single nucleotide polymorphisms, and insertions and deletions.

WO 2005/003390 A2